

ABSTRACT

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Title of diploma thesis:	Emerging treatment options for the mucopolysaccharidoses

This thesis provides a summary of knowledge about rare disease named mucopolysaccharidoses and about treatment options. Due to the low incidence in the population is in the common literature the disease mentioned only sporadically. The thesis is processed as a search, the primary method of work is collecting data by studying literature. The first part deals with general aspects of disease occurrence in the Czech Republic and abroad and mucopolysaccharides, in the second part are itemized in detail all known types of MPS, their clinical manifestations as well as a treatment that is currently quite problematic and unsatisfactory. In the treatment I focused mainly on the form of MPS III, because this form is the most widespread in the Czech Republic.

Key words: mucopolysaccharidosis, glycosaminoglycans, lysosomal storage disease, enzyme replacement therapy, substrate reduction therapy